Lipoid congenital adrenal hyperplasia with cholestatic jaundice

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Abstract

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BACKGROUND AND OBJECTIVE: Lipoid congenital adrenal hyperplasia (LCAH) is the most severe form of adrenal hyperplasia and mutations in the StAR gene are the most common cause of the disease. Adrenal insufficiency and cholestasis are reported in few patients. The aim of this study was to report the results of treatment of two sisters with lipoid CAH and cholestatic jaundice. CASE REPORTS: Here, we present two sisters at the age of 30 and 60 days with conjugated hyperbilirubinemia and elevated liver enzymes and adrenal insufficiency. They had a 46,XY karyotype with external female genitalia without uterus and ovaries. LCAH was detected through electrolyte abnormalities, increased ACTH, decreased levels of cortisol and sex hormones and was confirmed by determination of exome sequencing and Sanger sequencing. In these patients, a homozygous mutation (c.653C>T) in exon 6 of STAR gene was identified. The patients were treated with 10 mg of hydrocortisone IV every 8 hours for 3 days; oral hydrocortisone was then administered at a dose of 2.5 mg every 8 hours and 0.2 mg fludrocortisone daily. One month after the therapy, levels of bilirubin and liver enzymes of these patients became normal. The first patient died 7 months after her mother stopped giving the drugs to the child. The other patient is now 9 years old. She is in good clinical condition as her treatment goes on. CONCLUSION: Considering the reported cases, adrenal lipoid hyperplasia should be considered as a rare cause of cholestasis with adrenal insufficiency in patients.

Author keywords

Adrenal Insufficiency; Karyotype; Lipoid CAH; Neonatal Cholestasis